View metadata, citation and similar papers at core.ac.uk
brought to you by CORE
provided by Elsevier - Publisher Connector



## 400 Decreased bone mineral density in adolescents and young adults with cystic fibrosis: prevalence and risk factors

A. Jourdain[1], F. Labarthe[1], S. Willot[1], C. Giraut[1], F. Varaigne[2], S. Marchand[1], C. Maurage[1]. *[1]Paediatrics, CHRU, Tours, France; [2]Pneumology, CHRU, Tours, France*

**Aims:** To determine bone mineral density (BMD) status and potential risk factors of demineralization in adolescents and young adults with cystic fibrosis (CF).
**Methods:** Dual-energy X-ray absorptiometry (DEXA) was performed in a cohort of 33 CF patients (17.1±4.2 years) to measure BMD at the lumbar spine. Various clinical and biochemical parameters reflecting nutritional, respiratory, infectious and metabolic status were obtained at DEXA time and retrospectively in the prepubertal period (PPP). The data were analysed using univariate and multiple linear regressions.
**Results:** The median BMD Z-score was −1.14 SD [min −3.64; max 1.44]. Eleven patients (33%) had osteopenia (−2SD < Z-score ⩽ −1SD) and 8 (24%) had osteoporosis (Z-score ⩽−2 SD). The Z-score value was significantly related to the patient height (expressed in SD) in the PPP ($p < 0.01$) and at DEXA time ($p < 0.001$). Univariate linear regressions showed that Z-score value was significantly associated with parameters reflecting nutritional (caloric and calcium intake, BMI) and infectious (precipitins against *P. aeruginosa*) status during the PPP but not at the time of DEXA. Multiple linear regression ($R^2 = 0.37$) confirmed the association between Z-score and precipitins against *P. aeruginosa* ($p = 0.03$, β-coeff. = −1.08±0.47), BMI ($p = 0.05$, β-coeff. = 0.04±0.02) and caloric intake ($p = 0.05$, β-coeff. = −0.29±0.14) in PPP, whereas none of them at DEXA time was related to Z-score.
**Conclusion:** Decreased BMD is frequent (57%) among adolescents and young adults with CF, and related to prepubertal infectious and nutritional parameters. These results suggest that more effective strategies may be developed before puberty to prevent the bone demineralization during adulthood.

## 401 Effects of disease progression on peripheral muscle strength and bulk, aerobic capacity and physical activity in cystic fibrosis (CF)

C. Opdekamp, V. Dufresne, C. Knoop, M. Lamotte, G. Deboeck, B. Stallenberg, M. Estenne. *Chest Medicine, Erasme University Hospital, Brussels, Belgium*

Previous studies suggest that nutritional status and muscular function in CF patients worsen with disease progression, but how this deterioration is linked to the loss of lung function is not fully understood. The aim of this study was to characterize how progression of respiratory disease in CF patients impacts on nutritional status, peripheral muscular strength and bulk, aerobic capacity, and weekly physical activity (PA). We hypothesized that nutritional status and muscle function would follow a biphasic pattern, with values remaining relatively stable early on and then dropping when significant airflow obstruction develops.
We tested this hypothesis in 28 (16 males) clinically stable adult CF patients (age: 29±7 yrs; FEV1: 56±25%pred; BMI: 20.2±3.0 $kg/m^2$) and 17 (9 males) healthy controls (HC) matched for age and gender. Average (±SD) values in patients and HC were 374±114 vs 407±119N (NS) for isometric quadriceps strength (quadPT), 55±15 vs 60±13 $cm^2$ (NS) for quadriceps cross-section (quadCSA), 28.1±6.8 vs 34.4±7.5 $ml \cdot kg^{-1} \cdot min^{-1}$ ($p < 0.01$) for peak oxygen uptake ($pVO_2$), and 9±12 vs 17.8±18.6 $h \cdot w^{-1}$ (NS) for weekly PA assessed by Kriska's Modifiable Activity Questionnaire. In CF patients, the best fit between FEV1 and BMI, quadPT, quadCSA, $pVO_2$ and PA was obtained with linear correlations ($r = 0.44–0.68$, $p < 0.05$). In both groups, BMI was linearly correlated with quadPT and quadCSA ($r = 0.52–0.67$, $p < 0.05$). Furthermore, BMI was linearly correlated with PA in CF patients ($r = 0.62$, $p < 0.001$). In conclusion, the deterioration of muscle function, physical performance and nutritional status in CF patients parallels the loss of pulmonary function.
Supported by: Erasmus Foundation.

## 402 Lip enlargement as a clinical feature of cystic fibrosis

S.S. Cakic. *Clinic of Periodontology and Oral Medicine, Faculty of Stomatology, Belgrade, Yugoslavia*

Cystic fibrosis (CF) is a multisystemic life-threatening disorder with a dysfunction of exocrine glands as the main feature. It is inherited as an autosomal recessive trait, and therefore usually diagnosed early in infancy. Clinical features may significantly vary between patients, which could be the obstacle for early diagnosis of CF. Main sites of involvement are pulmonary and gastrointestinal tracts, liver, pancreas and sweat glands. Oral cavity is rather rarely involved. This may include lip enlargement (relatively common), and biofilm-induced enanthema of both free and attached gingiva associated with mild xerostomia (relatively uncommon).
We present the case of a man 32 years old, who reported to the specialist of periodontology and oral medicine due to chronic enlargement of a lower lip. Patient history did not reveal any general disease, except "frequently repeated respiratory infections". As 10-days topical application of corticosteroid ointment did not affect lip enlargement, we reffered the patient to his physician for further ivestigation. Two weeks later we were informed that CF was diagnosed in this patient. It is concluded that lip enlargement as a clinical feature of atypical CF should be kept in mind during diagnostic process.

## 403 Estimating the age of common *CFTR* mutations in Brittany (western France)

Y. Fichou[1,2], E. Génin[3], C. Le Maréchal[1,4], M.P. Audrézet[1,2], V. Scotet[1], C. Férec[1,4]. *[1]Inserm U613, Génétique Moléculaire et Génétique Epidémiologique, EFS – Région Bretagne, Brest, France; [2]Laboratoire de Génétique Moléculaire, CHRU Morvan, Brest, France; [3]Inserm U535, Génétique Epidémiologique et Structure des Populations Humaines, Hôpital Paul Brousse, Villejuif, France; [4]Génétique Somatique et Constitutionnelle, Université de Bretagne Occidentale, Brest, France*

Cystic fibrosis (CF) is the most common autosomal recessive disorder affecting children in Caucasian population. CF is caused by mutations within the CF transmembrane conductance regulator (*CFTR*) gene (7q31.2), which encodes a 1,480 aminoacid cAMP-dependent chloride channel. To date, more than 1,500 mutations and polymorphims within the *CFTR* gene have been reported. The p.F508del mutation accounts for ~70% of the total mutated alleles and only four additional mutations have a frequency >1% in the general population. However, disparities exist between specific populations depending on their ethnical and/or geographical origins. We used a simple likelihood-based method [Génin *et al.*, 2004] to estimate the age of the most recent common ancestors carrying *CFTR* mutations that are preferentially observed in Brittany (western France). The occurence of CF is particularly high in this region as compared with other areas: ~1/2,600 in Brittany *vs* ~1/3,500 in the Caucasian population, with a typical spectrum of mutations: $p.W846X_2$, c.1078delT, p.G551D and p.F508del mutations, as well as the p.I1027T polymorphism, a variant that is found in *cis* with the p.F508del mutation. By using our method, we were able to estimate the number of generations since each of the variants appeared in Brittany. This work actually provides historical perspectives in the field of understanding population migrations.